



General

Guideline Title

Essential elements of genetic cancer risk assessment, counseling, and testing: updated recommendations of the National Society of Genetic Counselors.

Bibliographic Source(s)

Riley BD, Culver JO, Skrzynia C, Senter LA, Peters JA, Costalas JW, Callif-Daley F, Grumet SC, Hunt KS, Nagy RS, McKinnon WC, Petrucelli NM, Bennett RL, Trepanier AM. Essential elements of genetic cancer risk assessment, counseling, and testing: updated recommendations of the National Society of Genetic Counselors. *J Genet Couns*. 2012 Apr;21(2):151-61. [78 references] [PubMed](#)

Guideline Status

This is the current release of the guideline.

This guideline updates a previous version: Trepanier A, Ahrens M, McKinnon W, Peters J, Stopfer J, Grumet SC, Manley S, Culver JO, Acton R, Larsen-Haidle J, Correia LA, Bennett R, Pettersen B, Ferlita TD, Costalas JW, Hunt K, Donlon S, Skrzynia C, Farrell C, Callif-Daley F, Vockley CW. Genetic cancer risk assessment and counseling: recommendations of the National Society of Genetic Counselors. *J Genet Counsel* 2004 Apr;13(2):83-114. [104 references]

Recommendations

Major Recommendations

Recommendation 1

The information collected at intake should include a thorough personal medical history and a 3-4 generation family medical history (pedigree), both of which are crucial for effective cancer risk assessment.

Recommendation 2

The genetic cancer risk assessment process should include using personal and family medical history information to determine whether an individual/family has an average, moderate or increased cancer risk. This information can then be used to generate a list of hereditary cancer syndromes to consider in the differential diagnoses.

Recommendation 3

Genetic testing should be offered when the following conditions apply:

- An individual has a personal or family history suggestive of an inherited cancer syndrome (American Society of Clinical Oncology [ASCO],

2003; Robson et al., 2010).

- The genetic test can be adequately interpreted (ASCO, 2003; Robson et al., 2010).
- Testing will influence medical management of the patient or other relatives (ASCO, 2003; Robson et al., 2010).
- The potential benefits of testing outweigh the potential risks.
- Testing is voluntary.
- The individual seeking testing or their legal proxy can provide informed consent.

Recommendation 4

An informed consent process is a necessary, and in some states legally required, component of genetic testing for hereditary cancer susceptibility and should precede genetic testing.

Elements of Informed Consent for Cancer Genetic Testing

1. Purpose of the test and who to test. An explanation should be provided that covers why the test is being offered, how the results might alter the individual's or their family members' cancer risk and the medical or surgical options to manage this risk. In the absence of an identified mutation in a family, the importance of first testing an affected relative should be discussed. If this is not an option, the limitations of testing an unaffected individual in the absence of an identified familial mutation should be described in detail with the patient (see below).
2. General information about the gene(s). Cancer risks associated with the gene mutation(s) in question, including the mode of inheritance and the concepts of genetic heterogeneity, incomplete penetrance and variable expressivity, should be reviewed.
3. Possible test results. The implications of all possible test results should be explained:
 - a. Positive Result (a deleterious mutation was identified): The identification of a known deleterious mutation guides management. In addition, it provides an opportunity for other at-risk relatives in the family (whether affected or unaffected with cancer) to undergo genetic counseling and testing for the identified familial mutation.
 - b. Negative Result (no deleterious mutations were identified):
 - i. Uninformative negative: In the absence of a known deleterious mutation in a family, a negative result is generally considered uninformative and must be interpreted with caution. It is important to stress the meaning of an uninformative test result to the patient, as failure to understand the significance of such a result may lead to the patient's nonadherence to recommended cancer screening or cancer risk reduction practices.
 - ii. True negative. If a patient tests negative for a known deleterious mutation that has been previously identified in a close biological relative, the patient is generally not considered to be at a significantly increased risk of developing cancer.
 - c. Variant of Uncertain Significance (VUS): In this case, a deoxyribonucleic acid (DNA) change in a gene has been identified, but it is unknown whether the DNA change affects gene function or if it represents normal variation. Genetic testing for the VUS should not be offered to other relatives on a clinical basis. Research studies involving the patient's relatives may help establish the clinical significance of the variant. Various online mutation evaluation programs may provide information to help interpret the significance of VUS (e.g., multivariate analysis of protein polymorphisms–mismatch repair [MAPP-MMR], <http://mappmmr.Blueankh.com/Impact.php> [redacted]; PolyPhen, <http://genetics.bwh.harvard.edu/pph/> [redacted]; Leiden Open Variation Database; BIC Database). The practitioner should be aware that there are notable limitations in the accuracy of such models and variant databases.
4. Technical aspects and accuracy of the test. The method(s) used for mutational analysis and the likelihood of a false-positive or false-negative result (sensitivity and specificity) should be reviewed (Eng, Hampel & de la Chapelle, 2001). The turn-around-time for results and method of disclosure should also be addressed.
5. Economic considerations. Patients should be apprised of the cost of genetic testing, and informed that their particular insurance plan(s) may not provide coverage or reimbursement for such tests.
6. Possibility of genetic information discrimination. Although there has been significant concern, there has been limited evidence of genetic discrimination in health insurance. Over the last two decades, new state and federal statutes in the United States have reduced the potential risk of genetic discrimination in health insurance and employment for a majority of citizens. Nonetheless, the status and limitations of legislation not only in health insurance and employment but also life, long-term, and disability insurance should be discussed with persons considering genetic testing for cancer predisposition.
7. Psychosocial aspects. A psychosocial assessment regarding testing should be performed. The assessment should include but not be limited to 1) anticipated reaction to results and coping strategies (Baum, Friedman & Zakowski, 1997; Croyle et al., 1997; Lerman et al., 1997); 2) timing and readiness for testing; 3) family issues; and 4) preparing for result disclosure. Failure to anticipate reactions accurately can lead to increased emotional distress months after testing (Dorval et al., 2000). See the original recommendations (Trepanier et al., 2004) for an expanded discussion of these aspects.
8. Confidentiality. Genetic counselors have an ethical responsibility and legal obligation to maintain patient confidentiality. Federal Health

Information Portability and Accountability Act (HIPAA) regulations require health providers to protect the privacy of all medical information, including genetic information. As a consequence, genetic counselors should share genetic testing results, pedigree and other medical information only as the patient directs (Gallo, Angst & Knafl, 2009). Patients should also be made aware whether the results will be disclosed to any third party (including the referring physician), and whether the center initiating the testing has any confidentiality safeguards. Several organizations have addressed the challenges of maintaining confidentiality in the genetic counseling setting (ASCO, 2003; Robson et al., 2010; "ASHG statement," 1998; Schneider et al., 2006). Because the results of genetic testing may be valuable to family members even if the patient is deceased, it is important to establish in advance which individual or individuals should have access to the patient's test results in their absence.

9. Utilization of test results. Options for cancer risk reduction and surveillance based on the patient's: level of risk for specific cancers, genotype, family history, medical history, dietary, and social habits should be presented. Patients should be encouraged to seek additional information from their referring physicians.
10. Alternatives to genetic testing. Patient decisions about genetic testing should be free from coercion. Patients may decide against genetic testing for cancer susceptibility for a variety of reasons including: 1) lack of interest, 2) indecision, 3) inadequate insurance coverage, and 4) uninformative results in an affected family member when genetic testing is not done, risk assessment and related recommendations should be made based on the family and medical history. When appropriate and feasible, an affected patient's DNA should be banked allowing designated at-risk family members the opportunity to seek genetic testing in the future (Brown, Moglia, & Grumet, 2006-2007; Quillin et al., 2010).

Recommendation 5

Disclosure of genetic test results should include personalized interpretation of results, cancer risk re-assessment, and identification of at-risk family members, regardless of whether the result is positive, negative, or inconclusive.

Recommendation 6

Psychosocial assessment is critical and should be part of both the pre-test and post-test genetic counseling process.

Clinical Algorithm(s)

None provided

Scope

Disease/Condition(s)

Hereditary cancer

Guideline Category

Counseling

Evaluation

Risk Assessment

Clinical Specialty

Family Practice

Internal Medicine

Medical Genetics

Obstetrics and Gynecology

Oncology

Preventive Medicine

Psychiatry

Intended Users

Advanced Practice Nurses

Nurses

Physician Assistants

Physicians

Psychologists/Non-physician Behavioral Health Clinicians

Social Workers

Guideline Objective(s)

To present a current and comprehensive set of practice recommendations for effective genetic cancer risk assessment, counseling, and testing

Target Population

Individuals who may benefit from cancer risk assessment, counseling, and genetic counseling

Interventions and Practices Considered

1. Personal medical history
2. Family medical history (3-4 generation)
3. Genetic cancer risk assessment
4. Genetic testing
5. Obtaining informed consent prior to genetic testing
6. Disclosure of genetic test results
 - Personalized interpretation of results
 - Cancer risk re-assessment
 - Identification of at risk family members
7. Psychological assessment

Major Outcomes Considered

- Sensitivity and specificity of cancer risk assessment tests
- Accurate assessment of cancer risk or risk of carrying a genetic mutation related to increased cancer risk

Methodology

Methods Used to Collect/Select the Evidence

Hand-searches of Published Literature (Primary Sources)

Description of Methods Used to Collect/Select the Evidence

The guideline authors searched via MEDLINE/PubMed, previously published guidelines and policy statements published by the American Society of Clinical Oncology, American Society of Human Genetics, American College of Medical Genetics, National Comprehensive Cancer Network, National Cancer Institute PDQ, United States Preventive Services Task force and the National Society of Genetic Counselors. The searches included English language, medical and psychosocial literature for the time period of 2004 to 2011. The search terms used were: cancer genetic counseling, cancer predisposition, hereditary cancer, informed consent, risk assessment, genetic susceptibility testing, family history, psychosocial assessment, and result disclosure.

Number of Source Documents

Not stated

Methods Used to Assess the Quality and Strength of the Evidence

Not stated

Rating Scheme for the Strength of the Evidence

Not applicable

Methods Used to Analyze the Evidence

Review

Description of the Methods Used to Analyze the Evidence

Not stated

Methods Used to Formulate the Recommendations

Expert Consensus

Description of Methods Used to Formulate the Recommendations

These guidelines were developed by the National Society of Genetic Counselors (NSGC) Familial Cancer Risk Counseling Special Interest Group which includes NSGC members providing cancer genetics services. Each practice guideline focuses on a clinical or practice-based issue, and is the result of a review and analysis of current professional literature believed to be reliable.

Rating Scheme for the Strength of the Recommendations

Not applicable

Cost Analysis

A formal cost analysis was not performed and published cost analyses were not reviewed.

Method of Guideline Validation

Not stated

Description of Method of Guideline Validation

Not stated

Evidence Supporting the Recommendations

References Supporting the Recommendations

American Society of Clinical Oncology. American Society of Clinical Oncology policy statement update: genetic testing for cancer susceptibility. *J Clin Oncol*. 2003 Jun 15;21(12):2397-406. [PubMed](#)

ASHG statement. Professional disclosure of familial genetic information. The American Society of Human Genetics Social Issues Subcommittee on Familial Disclosure. *Am J Hum Genet*. 1998 Feb;62(2):474-83. [PubMed](#)

Baum A, Friedman AL, Zakowski SG. Stress and genetic testing for disease risk. *Health Psychol*. 1997 Jan;16(1):8-19. [PubMed](#)

Brown KL, Moglia DM, Grumet S. Genetic counseling for breast cancer risk: general concepts, challenging themes and future directions. *Breast Dis*. 2006-2007;27:69-96. [129 references] [PubMed](#)

Croyle RT, Smith KR, Botkin JR, Baty B, Nash J. Psychological responses to BRCA1 mutation testing: preliminary findings. *Health Psychol*. 1997 Jan;16(1):63-72. [PubMed](#)

Dorval M, Patenaude AF, Schneider KA, Kieffer SA, DiGianni L, Kalkbrenner KJ, Bromberg JI, Basili LA, Calzone K, Stopfer J, Weber BL, Garber JE. Anticipated versus actual emotional reactions to disclosure of results of genetic tests for cancer susceptibility: findings from p53 and BRCA1 testing programs. *J Clin Oncol*. 2000 May;18(10):2135-42. [PubMed](#)

Eng C, Hampel H, de la Chapelle A. Genetic testing for cancer predisposition. *Annu Rev Med*. 2001;52:371-400. [75 references] [PubMed](#)

Gallo AM, Angst DB, Knafl KA. Disclosure of genetic information within families. *Am J Nurs*. 2009 Apr;109(4):65-9. [30 references] [PubMed](#)

Lerman C, Biesecker B, Benkendorf JL, Kerner J, Gomez-Caminero A, Hughes C, Reed MM. Controlled trial of pretest education approaches to enhance informed decision-making for BRCA1 gene testing. *J Natl Cancer Inst*. 1997 Jan 15;89(2):148-57. [PubMed](#)

Quillin JM, Bodurtha JN, Siminoff LA, Smith TJ. Exploring hereditary cancer among dying cancer patients--a cross-sectional study of hereditary risk and perceived awareness of DNA testing and banking. *J Genet Couns*. 2010 Oct;19(5):497-525. [PubMed](#)

Robson ME, Storm CD, Weitzel J, Wollins DS, Offit K, American Society of Clinical Oncology. American Society of Clinical Oncology policy statement update: genetic and genomic testing for cancer susceptibility. *J Clin Oncol*. 2010 Feb 10;28(5):893-901. [83 references] [PubMed](#)

Schneider KA, Chittenden AB, Branda KJ, Keenan MA, Joffe S, Patenaude AF, Reynolds H, Dent K, Eubanks S, Goldman J, Leroy B, Warren NS, Taylor K, Vockley CW, Garber JE. Ethical issues in cancer genetics: I 1) whose information is it. J Genet Couns. 2006 Dec;15(6):491-503. [PubMed](#)

Trepanier A, Ahrens M, McKinnon W, Peters J, Stopfer J, Grumet SC, Manley S, Culver JO, Acton R, Larsen-Haidle J, Correia LA, Bennett R, Pettersen B, Ferlita TD, Costalas JW, Hunt K, Donlon S, Skrzynia C, Farrell C, Callif-Daley F, Vockley CW. Genetic cancer risk assessment and counseling: recommendations of the National Society of Genetic Counselors. J Genet Couns. 2004 Apr;13(2):83-114. [104 references] [PubMed](#)

Type of Evidence Supporting the Recommendations

The type of supporting evidence is not specifically stated for each recommendation.

Benefits/Harms of Implementing the Guideline Recommendations

Potential Benefits

- The identification of patients with empirically increased risks allows for tailored screening and other interventions.
- The psychological impact of genetic testing amongst individuals who have never been affected by cancer demonstrates that many people with negative test results derive significant psychological benefits from genetic testing. One study suggests that cancer genetic risk assessment services help to reduce distress, improve the accuracy of the perceived risk of, and increase knowledge about, cancer and genetics.

Potential Harms

- Possibility of genetic information discrimination. Although there has been significant concern, there has been limited evidence of genetic discrimination in health insurance. Over the last two decades, new state and federal statutes in the United States have reduced the potential risk of genetic discrimination in health insurance and employment for a majority of citizens. Nonetheless, the status and limitations of legislation not only in health insurance and employment but also life, long-term, and disability insurance should be discussed with persons considering genetic testing for cancer predisposition.
- While no adverse effects have been observed among most people with positive test results, a select sub-set of people will be vulnerable to testing distress and require more professional assistance.

Qualifying Statements

Qualifying Statements

- The practice guidelines of the National Society of Genetic Counselors (NSGC) are developed by members of the NSGC to assist genetic counselors and other health care providers in making decisions about appropriate management of genetic concerns, including access to and/or delivery of services. Each practice guideline focuses on a clinical or practice-based issue, and is the result of a review and analysis of current professional literature believed to be reliable. As such, information and recommendations within the NSGC practice guidelines reflect the current scientific and clinical knowledge at the time of publication, are only current as of their publication date, and are subject to change without notice as advances emerge.
- In addition, variations in practice, which take into account the needs of the individual patient and the resources and limitations unique to the institution or type of practice, may warrant approaches, treatments and/or procedures that differ from the recommendations outlined in this guideline. Therefore, these recommendations should not be construed as dictating an exclusive course of management, nor does the use of such recommendations guarantee a particular outcome. Genetic counseling practice guidelines are never intended to displace a health care provider's best medical judgment based on the clinical circumstances of a particular patient or patient population. Practice guidelines are

published by NSGC for educational and informational purposes only, and NSGC does not "approve" or "endorse" any specific methods, practices, or sources of information.

Implementation of the Guideline

Description of Implementation Strategy

An implementation strategy was not provided.

Institute of Medicine (IOM) National Healthcare Quality Report Categories

IOM Care Need

Staying Healthy

IOM Domain

Effectiveness

Patient-centeredness

Identifying Information and Availability

Bibliographic Source(s)

Riley BD, Culver JO, Skrzynia C, Senter LA, Peters JA, Costalas JW, Callif-Daley F, Grumet SC, Hunt KS, Nagy RS, McKinnon WC, Petrucelli NM, Bennett RL, Trepanier AM. Essential elements of genetic cancer risk assessment, counseling, and testing: updated recommendations of the National Society of Genetic Counselors. J Genet Couns. 2012 Apr;21(2):151-61. [78 references] [PubMed](#)

Adaptation

Not applicable: The guideline was not adapted from another source.

Date Released

2004 Apr (revised 2012 Apr)

Guideline Developer(s)

National Society of Genetic Counselors - Medical Specialty Society

Source(s) of Funding

National Society of Genetic Counselors

Guideline Committee

Familial Cancer Risk Counseling Special Interest Group

Composition of Group That Authored the Guideline

Group Members: Bronson D. Riley, Julie O. Culver, Cecile Skrzynia, Leigha A. Senter, June A. Peters, Josephine W. Costalas, Faith Callif-Daley, Sherry C. Grumet, Katherine S. Hunt, Rebecca S. Nagy, Wendy C. McKinnon, Nancie M. Petrucelli, Robin L. Bennett, Angela M. Trepanier

Financial Disclosures/Conflicts of Interest

Not stated

Guideline Status

This is the current release of the guideline.

This guideline updates a previous version: Trepanier A, Ahrens M, McKinnon W, Peters J, Stopfer J, Grumet SC, Manley S, Culver JO, Acton R, Larsen-Haidle J, Correia LA, Bennett R, Pettersen B, Ferlita TD, Costalas JW, Hunt K, Donlon S, Skrzynia C, Farrell C, Callif-Daley F, Vockley CW. Genetic cancer risk assessment and counseling: recommendations of the National Society of Genetic Counselors. *J Genet Counsel* 2004 Apr;13(2):83-114. [104 references]

Guideline Availability

Electronic copies: Available to subscribers from the [Journal of Genetic Counseling Web site](#) .

Availability of Companion Documents

The following is available:

- Bennett RL, French KS, Resta RG, Doyle DL. Standardized human pedigree nomenclature: update and assessment of the recommendations of the National Society of Genetic Counselors. *J Genet Couns* 2008 Oct;17(5):424-33. Available to subscribers from the [Journal of Genetic Counseling Web site](#) .

Patient Resources

None available

NGC Status

This NGC summary was completed by ECRI Institute on May 11, 2012. The information was verified by the guideline developer on May 30, 2012.

Copyright Statement

This NGC summary is based on the original guideline, which is subject to the guideline developer's copyright restrictions.

Disclaimer

NGC Disclaimer

The National Guideline Clearinghouse[®] (NGC) does not develop, produce, approve, or endorse the guidelines represented on this site.

All guidelines summarized by NGC and hosted on our site are produced under the auspices of medical specialty societies, relevant professional associations, public or private organizations, other government agencies, health care organizations or plans, and similar entities.

Guidelines represented on the NGC Web site are submitted by guideline developers, and are screened solely to determine that they meet the NGC Inclusion Criteria which may be found at <http://www.guideline.gov/about/inclusion-criteria.aspx>.

NGC, AHRQ, and its contractor ECRI Institute make no warranties concerning the content or clinical efficacy or effectiveness of the clinical practice guidelines and related materials represented on this site. Moreover, the views and opinions of developers or authors of guidelines represented on this site do not necessarily state or reflect those of NGC, AHRQ, or its contractor ECRI Institute, and inclusion or hosting of guidelines in NGC may not be used for advertising or commercial endorsement purposes.

Readers with questions regarding guideline content are directed to contact the guideline developer.